Gaucher disease in South Africa

To the Editor: I refer to the abstract by Govender and Newton in the August *SAMJ*.[1]

Gaucher disease is a relentless progressive multi-systemic disorder caused by deficiency or inadequate function of lysosomal β-glucocerebrosidase. The resultant accumulation of the substrate glucocerebroside causes the organ damage. The classic clinical picture of organomegaly, cytopenia and bone pain or disease should always alert the practitioner and place Gaucher disease into the differential diagnosis. This will result in earlier intervention and minimise the risk of irreversible complications of the disease.

While initially Gaucher disease was thought to be more prevalent in the Jewish Ashkenazi population, it is now regarded as being pan-ethnic with a specific genotype, the N370S mutation being more prevalent in that group. There are now some 300 mutations causing the disease.

The Gaucher Clinic at Johannesburg Hospital has been in existence for some 14 years, and 30% of the patients are black Africans. It is interesting to note that there does appear to be a novel mutation for our black population. There are about 50 known patients in South Africa. The treatment of choice is

enzyme replacement therapy with imiglucerase (Cerezyme), and at present there are about 30 patients on this programme. An alternative form of therapy is substrate reduction; medication with miglustat (Zavesca) reduces the amount of glucocerebroside, allowing the patient's depleted residual glucocerebrosidase activity to cope with the reduced amount of substrate. There are at present 5 patients on substrate reduction therapy in South Africa. Both modalities of therapy are efficacious.

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1. Govender K, Newton KA. Gaucher disease in an African teenager (Abstract). *S Afr Med J* 2007; 97: 618.